Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by cutaneous mastocytosis, microcephaly, microtia and/or hearing loss, hypotonia and skeletal anomalies (e.g. clinodactyly, camptodactyly, scoliosis). Additional common features are short stature, intellectual disability and difficulties. Facial dysmorphism may include upslanted palpebral fissures, highly arched palate and micrognathia. Rarely, seizures and asymmetrically small feet have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mastocytosis-short stature-deafness syndrome
Mastocytoza - niski wzrost - utrata słuchu
Mastocytosis-short stature-hearing loss syndrome

Kod ORPHA
2135

Kod OMIM
248910

Kod ICD10
Q82.2

Kod ICD11
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*Źródło

orphanet